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Digital health: Patient-generated health data and use of digital technology

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ABBREVIATIONS

AI - Artificial Intelligence

EHR - Electronic Health Record

ISPUP - Instituto de Saúde Pública da Universidade do Porto (Institute of Public Health of the University of Porto)

IT - Health Information Technology

LTHCs - Long-Term Health Conditions

mHealth - Mobile Health

PGHD - Patient-Generated Health Data

PROs - Patient-Reported Outcomes

UHCSJ - Unidade Hospitalar do Centro de Saúde de São João (São João Health Center Hospital Unit)

WHO - World Health Organization

RESUMO

A digitalização e a criação de novas tecnologias aumentaram significativamente na área da saúde. As tecnologias digitais tornaram-se ferramentas importantes na relação entre pacientes, médicos e o sistema de saúde. Essas tecnologias permitem que os pacientes utilizem dispositivos digitais para monitorar sua saúde, compartilhem esses dados com suas equipes de saúde e facilitem a tomada de decisões sobre tratamentos. Os Dados de Saúde Gerados por Pacientes (*Patient-Generated Health Data*, PGHD) referem-se a informações relacionadas à saúde produzidas, documentadas ou coletadas por pacientes, familiares ou outros cuidadores informais para lidar com questões de saúde. Os PGHD abrangem vários aspectos, incluindo, mas não se limitando a, histórico de saúde, histórico de tratamento, sintomas, dados biométricos e medidas de resultados relatados pelos pacientes. Os PGHD estão desempenhando um papel crucial na melhoria da eficiência operacional dos sistemas de saúde. Ao capacitar pacientes com doenças raras ou seus cuidadores informais a coletar e monitorar dados relacionados à saúde, eles podem atuar como defensores e pesquisadores de sua condição. Devido à especificidade e complexidade dessas doenças, em um campo de conhecimento em constante evolução, o compartilhamento de dados de saúde gerados pelos próprios pacientes com prestadores de cuidados ou outros interessados pode ajudar a aprimorar a pesquisa científica e melhorar os benefícios clínicos. Esta tese busca explorar a disposição e as motivações dos pacientes com doenças raras e seus cuidadores informais para coletar ativamente seus dados de saúde e identificar as tecnologias preferidas para coletar esses dados. Além disso, também visa entender se os pacientes com doenças raras e seus cuidadores informais estão dispostos a compartilhar essas informações e para quais finalidades. Para alcançar esses objetivos, foi realizado um estudo qualitativo, e entrevistas semiestruturadas foram conduzidas, tanto por telefone quanto pessoalmente, com uma amostra de 24 pacientes e 16 cuidadores informais. A análise preliminar dos dados das transcrições das entrevistas foi realizada simultaneamente à coleta de dados para identificar prontamente informações

redundantes e, assim, interromper a amostragem quando necessário, utilizando o software NVivo V12. Posteriormente, foi realizada uma análise aprofundada focada nos principais tópicos da tese, utilizando abordagens dedutivas e indutivas. O estudo revelou que a maioria dos pacientes com doenças raras e seus cuidadores estão dispostos a coletar dados de saúde, com motivações diferentes entre os dois grupos. Para os pacientes, o principal incentivo era a possibilidade de usar os dados para melhorar o tratamento e a gestão da doença, enquanto os cuidadores informais estavam mais motivados pela oportunidade de contribuir para a pesquisa científica. A maioria dos participantes preferiu usar dispositivos móveis para a coleta de dados, embora alguns tenham optado por métodos analógicos. Em relação ao compartilhamento de dados, a maioria concordou em fornecer informações coletadas para pesquisas biomédicas e medicina personalizada, mas levantou preocupações sobre privacidade e segurança. Além disso, houve relutância em compartilhar dados para outros estudos, dependendo do propósito da pesquisa. A tese destaca a importância de compreender as diferentes motivações e preferências, bem como as preocupações, em relação à auto-geração e compartilhamento de dados de saúde, e conclui que, ao criar soluções para enfrentar essas barreiras identificadas, os sistemas de saúde, o tratamento dos pacientes e os estudos científicos sobre essas doenças podem se beneficiar do volume desses dados.

Palavras-chave: compartilhamento de dados: digitalização na saúde; dados de Saúde Gerados por Pacientes (PGHD); doenças raras; tecnologias digitais.

ABSTRACT

The digitization and creation of new technologies have significantly increased in healthcare. Digital technologies have become important tools in the relationship between patients, doctors, and the healthcare system. These technologies allow patients to use digital devices to monitor their health, share this data with their healthcare teams, and facilitate decision-making about treatments. Patient-Generated Health Data (PGHD) refers to health-related information produced, documented, or collected by patients, family members, or other informal carers to address health issues. PGHD encompasses various aspects, including but not limited to, health history, treatment history, symptoms, biometric data, and patient-reported outcome measures. PGHD is playing a crucial role in improving the operational efficiency of healthcare systems. By empowering patients with rare diseases or their informal carers to collect and monitor health-related data, they can act as advocates and researchers for their condition. Due to the specificity and complexity of these diseases, in a field of knowledge that is constantly evolving, sharing self-generated health data with providers or other stakeholders may help enhance scientific research and improve clinical benefits. This thesis seeks to explore the willingness and motivations of patients with rare diseases and their informal carers to actively collect their health data and identify their preferred technologies to self-generate data for this task. In addition, it also aims to understand whether rare disease patients and informal carers are willing to share this information and for what purposes. To achieve these goals, a qualitative study was undertaken, and semi-structured interviews were conducted, both by phone and in person, with a sample of 24 patients and 16 informal carers. Preliminary data analysis of interview transcripts was conducted concurrently with data collection to promptly identify redundant information and subsequently stop sampling as necessary using the NVivo software V12. Subsequently, an in-depth analysis focused on the key topics of the thesis was performed, utilizing both deductive and inductive approaches. The study revealed that most rare disease patients and their caregivers are willing to collect health data, with differing motivations between the two groups. For patients, the main incentive was the potential to use the data to improve treatment and disease management, while informal caregivers were more motivated by the opportunity

to contribute to scientific research. Most participants preferred using mobile devices for data collection, although some opted for analog methods. Regarding data sharing, the majority agreed to provide self-generated information for biomedical research and personalized medicine but raised concerns about privacy and security. Additionally, there was reluctance to share data for other studies, depending on the research purpose. The thesis highlights the importance of understanding the different motivations and preferences, as well as the concerns, regarding the self-generation and sharing of health data and concludes that by creating solutions to address these identified barriers, healthcare systems, patient treatment, and scientific studies on these diseases can benefit from the volume of this data.

Keywords: data sharing; digitalization in healthcare; digital technologies Patient-Generated Health Data (PGHD); rare diseases.

1. INTRODUCTION

Digitization and the creation of new technologies have increased in the healthcare field (Vayena, 2018). Digital technologies have become a major device in the relationship between patients, doctors, and the healthcare system. By connecting and promoting access to relevant health information, patients are being encouraged to use digital devices during their treatment journey, share this data with their healthcare team, and facilitate decision-making about treatment (Adler-Milstein & Nong, 2019; Cohen et al., 2016).

One of the challenges of disease management and treatment, especially for patients with long-term health conditions (LTHCs), is ensuring adherence and the high costs to the healthcare system. The use of digital devices, such as apps and wearables for monitoring and collecting health data generated by the patient, could be one of the solutions for increasing adherence to treatment and reducing costs (Brown R, 2022; Chen, 2020).

Patient-Generated Health Data (PGHD) refers to health-related information produced, documented, or gathered by patients, family members, or other carers to address health issues. PGHD encompasses various aspects, including, but not limited to, health history, treatment history, symptoms, biometric data, and patient-reported outcome measures (Calegari & Fettermann, 2022; ONC). PGHD is increasingly playing a crucial role in enhancing the operational efficiency of healthcare systems (Psiha, 2017). Using mobile apps and wearables, PGHD enables individuals' health to be tracked and monitored by patients and health professionals, including self-reported health and treatment histories and patient-reported outcomes (PROs), allowing them to capture relevant health information, monitor symptoms, and increase self-awareness about their medical conditions. Furthermore, they can serve as powerful devices in delivering safe, efficient, and patient-centered care, making healthcare provision more personalized and effective (Lavallee et al., 2020; Omoloja, 2021).

Despite the advantages identified, there are many barriers and challenges to using PGHD, such as ease of access to the necessary technology, privacy,

security, and confidentiality of the data. It has also been found that the amount of data collected can overwhelm patients and that they may have difficulty interpreting the data generated and experience negative feelings by constantly remembering the disease (Brown et al., 2022; Chen, 2020; Psiha, 2017; Rathbone et al., 2023).

In the context of patients with rare diseases, whose conditions often present unique and complex challenges and a life-long presentation, the adoption of PGHD assumes even greater importance. By empowering these patients to collect and monitor health-related data, they can play the role of advocates and researchers of their condition (Chen, 2020). Due to the specificity and complexity of these diseases, within a constantly evolving field of knowledge, sharing health data with their providers or other stakeholders can be a way to help enhance scientific research and improve clinical benefits (Courbier et al., 2019; de Freitas et al., 2017; Francisco et al., 2022). However, despite growing interest in this field, there is still limited qualitative research on the perspectives of patients with chronic conditions, especially patients with rare diseases, and their informal carers, particularly concerning perceived facilitators and concerns with generating and sharing their data. Currently, most studies on patient-generated health data collection are quantitative and there is limited research on the perspectives and subjective experiences of patients and their informal carers. Considering this, the main objective of this thesis is to explore the perspectives of people affected by rare diseases about engagement with health data self-generation, to identify the technologies (digital or analogic) they prefer to use for this task and to understand whether they would share that information and for which purposes.

In this chapter, I draw on existing literature to define the concepts of digital health and patient-generated health data while delving into the motivations and obstacles linked to collecting one's health information. Subsequently, I review the empirical studies concerned with patients' perspectives on sharing self-generated data and the purposes they can be used.

1.1. Digital health

1.1.1. Digital health technology

Technological advancements have fundamentally transformed healthcare, ushering in dramatic changes over the years (Thimbleby, 2013). Several hospitals and health systems have adopted digital technologies in various functional domains, such as implementing electronic health record (EHR) systems, developing apps, and experimenting with disruptive technologies such as artificial intelligence (AI) (Stoumpos et al., 2023).

Digital health covers the use of information and communication technologies in medicine and various health professions and includes mobile health (mHealth), health information technology (IT), wearable devices, telehealth and telemedicine, and personalized medicine. It aims to reduce health risks, manage diseases, and promote general well-being (Ronquillo et al., 2023; U.S. Food & Drug Administration, 2020). The utilization of digital devices enables the digitization, automatic classification, structuring, and presentation of health data. This information can be displayed in meaningful and actionable formats, such as dashboards, graphs, and figures, offering real-time insights. Additionally, data can be collected daily, providing a comprehensive view of a patient's ongoing health status for the health providers (Jen et al., 2023).

According to the World Health Organization (WHO) (2018), the dissemination of digital technologies and global interconnectivity holds substantial potential to expedite countries' advancements in achieving universal health coverage and ensuring guaranteed access to high-quality health services. In the past, health records were mostly on paper, with the result that data was often uninterpretable, illegible, and incomplete, and therefore the analysis of this information was limited. Increasing the implementation of digital health, especially mHealth, is crucial for countries to offer access to high-quality healthcare (Jen et al., 2023; World Health Organization, 2018).

Many studies cite the advantages of using wearable devices and mHealth applications and other eHealth technologies, for example: their significant

potential to improve access to healthcare for a diverse range of social groups through remote interventions, namely by offering alternatives to face-to-face consultations for individuals experiencing health-related mobility limitations, the elderly, and people residing in locations geographically distant from medical centers; as well as an additional avenue to meet the health needs of those who lack access to quality medical information and care, health monitoring and the promotion of positive health habits, among others (Brown et al., 2022; Calegari & Fettermann, 2022; Wu DTY, 2020).

Recent research has indicated that the acceptance of emerging mobile technologies, such as health apps, is significantly influenced by factors like socioeconomic status, age, and digital literacy. On a patient level, challenges include issues related to data collection, access to the internet, apprehensions regarding data security and privacy, particularly the potential misuse of health information, and concerns about facing judgment based on their health condition (Brown et al., 2022; Fiske, Prainsack, et al., 2019; Omoloja, 2021; Rathbone et al., 2023). Other obstacles observed were that self-registration devices lack flexibility and standardized formats, which makes it difficult for the health team to interpret this data, ensure consistent use, and prevent the abandonment of the devices, and that the need to collect one's health data can contribute to negative feelings of constantly remembering one's illness (Chen, 2020; West et al., 2017).

1.1.2. Patient-generated health data: benefits and barriers

Growing public interest in health matters has spurred the creation of health-focused technological products and personal health data generation. PGHD plays a pivotal role in tackling health issues by providing a thorough depiction of patients' health conditions. It distinguishes itself from data produced in clinical settings, and during interactions with healthcare providers, in two crucial ways. Firstly, the primary responsibility for capturing or documenting this data lies with patients, and not with healthcare providers. Secondly, patients have the authority to determine how they wish to share or disseminate this information with providers and other relevant parties. Examples of PGHD include blood pressure readings using home health equipment and glucose monitoring and diet

recording using a mobile app, among others (Office of the National Coordinator for Health Information Technology, 2014; Omoloja, 2021).

The primary advantages of employing PGHD include the capability for ongoing information gathering, accessibility outside traditional healthcare environments, patient self-management, insights into both patient health and behaviors, the revelation of unexpected side effects, the facilitation of timely and cost-effective interventions, and the vital support it offers for the continuity of care and patient (Chen, 2020; Lavallee et al., 2020; Öberg et al., 2018). Another benefit associated with PGHD is personalized, efficient, and collaborative care: due to the health data generated, doctors and the healthcare team can have the opportunity to obtain information that can justify symptoms and health outcomes and, in turn, help with decision-making about the course of treatment and in providing additional support when needed (Cerna, 2020; Chen, 2020; Kawu et al., 2023).

Despite the benefits identified, there are numerous barriers to the use of PGHD. At the provider level, there are concerns about the quality of information coming from self-monitoring practices, such as the reliability and validity of self-tracking devices, as they may be considered less reliable than clinical laboratory data and, in some cases, unreliable (Piwek et al., 2016; West et al., 2017). Other challenges identified in the literature encompass the risk of participants manipulating their health data, thereby providing an inaccurate portrayal of their health condition. Additionally, the brevity of consultation time may lead healthcare providers to expend a considerable amount of time deciphering the collected data, potentially compromising the quality of patient care. The lack of data integration in the electronic medical record was also identified as an additional challenge (Chen, 2020; Jim et al., 2020).

In addition, physicians may find it difficult to analyze PGHD due to a lack of time and resources. As medical care is changing and becoming more data-driven, and there is a lack of specific training for doctors in patient-generated data analysis, it has been suggested that a new professional specialty – health information counselors, should be created. These professionals would specialize in medical data science, hold analytical skills in statistics and data interpretation, and work at healthcare establishments to support both patients and healthcare

professionals in discerning between the advantages and pitfalls of the use of digital or commercial health information sources when making healthcare decisions (Fiske, Buyx, et al., 2019). Preliminary findings show that rare disease patients and their informal carers can benefit from the support of this new set of professionals (De Freitas et al., 2024).

1.1.2.1. Motivations for self-generating and sharing data for biomedical research and personalized medicine

The imperative to share health data for the advancement of biomedical research and enhancement of clinical outcomes is well-documented, especially in the realm of rare diseases. This domain faces unique challenges due to limited knowledge and expertise, coupled with geographically dispersed patient populations (Courbier et al., 2019; Modi et al., 2023). In the field of biomedical research, the motivations for sharing health data are driven by the potential to deepen our understanding of diseases, discover cures for untreatable illnesses, develop new drugs, engage in meaningful discussions, and gain insights directly from the professionals involved in the projects, if their health data is shared anonymously regardless of the severity of their disease and their sociodemographic profile (Amorim et al., 2022; Brown et al., 2022; Courbier et al., 2019; Rathbone et al., 2023).

The question of who will be granted access to this data holds significant importance. According to a study by McCormark and colleagues (2016), most participants expressed a preference for restricting access to their records exclusively to individuals operating within the realm of health and health research. Furthermore, unanimous consensus among all participants is observed in their opposition to data access by private entities, encompassing insurance, financial services, and healthcare providers. Their concern lies in the apprehension of potential discrimination if access to the data is not meticulously regulated.

Another significant incentive for self-generating health data is personalized medicine. Due to the volume of data generated from digital devices, this data can serve as a valuable resource for providers to recognize and predict patterns, allowing them to offer personalized care and insights into patients' health status

and conditions, as well as the unique biological, social and environmental factors that affect them, in addition to simplifying the diagnostic process (Cortez, 2018; Loos & Davidson, 2016; Zhu et al., 2016).

The notion of collaborative decision-making is also emphasized in this context. Adopting a personalized approach to care planning involves open discussions between patients and healthcare professionals regarding the issues that hold the utmost importance for the patients. This approach aims to align care with the unique needs and priorities of everyone. Patients are actively encouraged to establish recovery goals that go beyond symptomatology, encompassing other aspects of their lives that contribute to overall well-being, such as behavioral changes, the development of new skills, social roles or activities for physical exercise, and personal challenges to overcome (Menear et al., 2022).

1.2. Use and impacts of patient-generated health data

In the last 10 years, there has been a notable increase in empirical studies exploring digital health, with specific emphasis on the use of PGHD and its impacts. This sub-section provides a review of the literature and key findings on these topics, such as the integration of patient-produced data with electronic health reports, the importance of usability and user experience in building a patient application, the impacts that collecting this data can have on disease management and data sharing benefits and challenges.

1.2.1. Patient-generated health data integration with Electronic Health Records (EHR)

Numerous studies have explored the integration of PGHD into Electronic Health Records (EHR) (Lobach et al., 2022; Shaw et al., 2022; Zhang et al., 2019). Key findings from this body of research underscore its positive impact, including enhanced care coordination, improved clinical decision-making, and heightened patient engagement. Nevertheless, persistent methodological

challenges, such as standardization issues and interoperability concerns, pose significant obstacles. The central question revolves around striking a delicate balance between data standardization and the flexibility necessary to accommodate the diverse sources of PGHD.

The integration of this data to support clinical care has been observed in several studies (Kumar et al., 2016; Shaw et al., 2022; Zhu et al., 2016). The benefits described are the possibility of improving patient outcomes, coordination of care, quality, and cost-effectiveness in ambulatory settings. For example, simplifying information processing tasks and avoiding duplicating diagnostic or therapeutic interventions, helps to reduce costs and improve care. It also facilitates another model of care, such as telemedicine, which, by integrating this data, enables remote medical interventions, helps to reduce waiting queues, and increases the flexibility of care (Bokolo Anthony, 2020; Calegari & Fettermann, 2022; Wicks et al., 2014).

However, other studies (Antonio et al., 2022; Zhang et al., 2019) have revealed technical challenges, such as a lack of operability between systems and logistical, social, and organizational obstacles, when incorporating patient-reported outcomes into clinical care. These obstacles result in heavy task loads, disruptions to clinical workflows, and the absence of a display of longitudinal data. To overcome these complex barriers, innovative and collaborative strategies are essential, such as facilitating, standardizing, and optimizing data collection, improving data visualizations, and ensuring user understanding (Kathrin et al., 2018; Zhang et al., 2019).

1.2.2. Digital health applications for self-generating health data: usability and user experience

Studies investigating the usability of digital health applications highlight the importance of user-friendly interfaces (Brown et al., 2022; Rahdar et al., 2023; Wu DTY, 2020). Currently, several devices and applications track and monitor health and well-being, including wearable devices. Some of these applications automatically monitor heart rate, steps taken, calories burned, and blood pressure, whereas in others these data are manually added by people, such as

diet records, water intake, weight, blood glucose, and lifestyle behavior (Brown et al., 2022; Lv et al., 2017; Öberg et al., 2018; Papa et al., 2020).

The measurements provided by the applications are not only intended to impact and help the user or carers to monitor some behavior or disease, but also, if applicable, to support the health professionals who are caring for them, providing health information outside the hospital environment. To achieve the application's objective, it must be developed in a user-friendly way (Hesse & Shneiderman, 2007).

Numerous studies suggest that app design is linked to patient engagement and the promotion of healthy habits. Strategies such as creating challenges, feedback, and rewards can increase engagement, self-tracking, and motivation. In addition, the ease of use of the device is an important factor, especially for patients with physical restrictions or dependent on carers, and it is not considered time-consuming, useless, or inconvenient (Patrick et al., 2019; Peng et al., 2016; Stiglbauer et al., 2019). However, obstacles such as limited access to digital devices and a lack of knowledge on how to capture and report health data can hinder users' engagement with the digital technologies (Adler-Milstein & Nong, 2019; Peng et al., 2016; Wu DTY, 2020).

Due to the difficulties that users can present, such as capturing information incorrectly or misinterpreting data, they must be provided with digital health training to increase the digital literacy of patients, carers, and health professionals. In addition, applications must be developed in a patient-centered way, aligned with the needs and reality of each patient (Hesse & Shneiderman, 2007; Kathrin et al., 2018; Slevin et al., 2019).

1.2.3. Impact of patient-generated health data on disease management, patient engagement, and behavior change

Many studies have shown significant impacts of PGHD on disease management, patient engagement, and behavior change. These findings suggest the potential of PGHD to empower individuals to manage their health and improve communication between patients and providers (Loiselle & Ahmed, 2017; Peng

et al., 2016). However, there are some barriers, such as privacy and security issues and device abandonment (Ibid).

Expected benefits of PGHD include increasing the knowledge of the patient and carers about the disease, the possibility to share with the provider relevant information about their diagnosis and management of the disease, and preventive care actions (Adler-Milstein & Nong, 2019; Patrick et al., 2019). The ability of digital solutions to modify and personalize the way people interact with health facilities was also described. This remote monitoring can also reduce hospitalization costs, since most critical cases will likely receive attention earlier, identifying changes in disease activity and symptoms in real-time and offering appropriate support promptly, making patients and their carers more engaged in their consultations and treatment (Austin et al., 2020; Öberg et al., 2018; Papa et al., 2020; Shen et al., 2022). The drive to use applications can also be related to the rewards created by the app itself, stimulating a behavior change. These rewards are closely linked to apps related to physical activity and dieting (Peng et al., 2016).

Despite the many advantages associated with self-generating health data, some studies report that patients consider the benefit of self-monitoring to be temporary, and many of them abandon it within a few months. This is due to several factors, such as the change in behavior having already been made, learning about the disease and routine having been obtained, where the usefulness was only considered to initially assess the disease and its routine (Peng et al., 2016; Stiglbauer et al., 2019).

1.2.4. Sharing self-generated health data: purposes, benefits, and challenges

Self-generated health data can be shared for research, as well as to support the choice of specific treatment models by the medical team in charge, or for enabling patients and their families and friends to make decisions. Several authors have studied the benefits and risks of sharing data (Brown et al., 2022; Fiks et al., 2014; Sigurðardóttir, 2022; Wicks et al., 2012).

One of the benefits identified by the literature is the possibility for patients to understand their state of health and symptoms in relation to other patients with the same disease, and even to receive updates on research and community events. In addition, it has also been observed that sharing information with family members or friends can provide informational and emotional support when they have to make challenging decisions (Peng et al., 2016; Wicks et al., 2012). In the group of patients who were already accustomed to sharing information with professionals, sharing self-generated health data with health professionals was also considered to be “easy” and “supportive”, one of the reasons being the understanding that such sharing would be beneficial to their treatment. (Sigurðardóttir, 2022).

Data sharing is a very sensitive topic, due to privacy and security issues (Loiselle & Ahmed, 2017). For some patients, sharing with others is perceived as meaningless and considered a private concern. Other patients do not feel comfortable sharing with friends or family, only with those with whom it makes sense to share. A study by Brown and colleagues (2020) shows that 54% of participants believe that sharing their health data would make people act differently towards them. Another concern is the misuse of this data, whether by health companies, health insurers, or advertisers, whom participants feel can put them in danger if they get access to those data (e.g. denying health insurance due to knowledge about the presence of a pre-existing health condition) (Peng et al., 2016).

Data leakage has also been discussed as a chief concern. For some diseases, the leakage of this data can cause people suffering from particular diseases to be stigmatized and damage their social relationships. Sharing data can be positive if the patient and/or their carers are sure about who and how they will use it. If the issues of security and privacy are overcome, more patients may be willing to share their health data (Brown et al., 2022; McCormack et al., 2016).

2. OBJECTIVES

The primary aim of this study lies in gaining a comprehensive understanding of patients' and informal carers' perspectives about active engagement in the generation and sharing of patient health data, by inquiring people affected by rare diseases. Its specific objectives are threefold:

1. To explore the willingness of rare disease patients and informal carers to self-generate health data and their motivations and concerns
2. To understand participants' preferences concerning the use of digital and analog technologies to self-generate health data and their underlying reasons.
3. To examine participants' willingness to share self-generated health data for personalized medicine, biomedical research, and other purposes, as well as their motivations and concerns.

3. METHODS AND ANALYSIS

3.1. Study design

This study draws on semi-structured qualitative interviews conducted within the scope of a larger mixed-methods research project addressing public and patient involvement in health data governance and titled DATAGov, whose protocol is available elsewhere (de Freitas et al., 2021). Specifically, this study focuses on patients' and informal carers' willingness to self-generate and share patient health data, as well as on participants' preferences and concerns about the use of digital technologies to self-generate that data.

The qualitative study undertaken under the DATAGov project was preceded by a hospital-based survey conducted at two reference centers for rare diseases at the Local Health Unit São João (ULSSJ)[1]: the Reference Centre for Inherited Metabolic Disorders and the Reference Centre for Congenital Heart Diseases. Participant recruitment for the survey included all patients aged 12 and above with a diagnosis of inherited metabolic disorders or congenital heart diseases or were in the process of being diagnosed, and their informal carers, mostly family members. Participants were informed about the study by a health professional and invited to take part in the survey by a research team member. For the interviews, only adult participants were recruited: all patients aged 18 or older, and their informal carers, who gave consent and completed the survey were subsequently asked permission by writing to be contacted within approximately four months by a researcher to invite them to participate in a semi-structured interview. The DATAGov project protocol, within which this study was carried out, received ethical approval from the Ethics Committee for Health from the UHCSJ/Faculty of Medicine of the University of Porto (Ref. 99/19).

[1] This service has recently changed its name from University Hospital Centre São João (UHCSJ).

3.2. Sampling and participants

From the 544 participants eligible for a semi-structured interview, 508 accepted to be contacted, including 60 participants and 448 informal carers (93.4% acceptance rate). Participants residing in Porto, Braga, and Aveiro districts were purposefully selected to participate in the interview and contacted by telephone or email by a researcher, according to their expressed preference. Between July 2019 and March 2020, 90 participants (83 informal carers and 7 patients) were contacted and 72 refused participation in the interview (66 informal carers and 6 patients): 41 did not answer the phone or email after several contacts, 28 were unavailable to do the interview and three had no interest to participate in the study. In total, 18 participants participated in in-person interviews (17 with carers and 1 with a patient).

In March 2020, the Portuguese government declared a state of emergency due to the COVID-19 pandemic and recruitment was stopped. To avoid causing any further health complications to participants through unnecessary exposure, data collection protocols and instruments were re-adjusted by the research team to carry out the interviews by telephone. Between August and October 2020, 43 patients were contacted by phone or e-mail, following their preference, to participate in a telephone interview and 19 refused to participate: 14 did not answer the phone or e-mail after several contacts, and five were unavailable to do the interview. In total, 23 patients participated in telephone interviews.

3.3. Data collection and analysis

Interviews were held at the location and time preferred by participants. In the case of informal carers, 17 semi-structured interviews were conducted in person at the university department responsible for the study (n=6), in a venue close to participants' homes or workplaces (n=9) and at participants' homes (n=2). Interview duration ranged from 20 to 53 minutes (Mean: 36 minutes). As for patients, 23 interviews were conducted by telephone and 1 interview was conducted in-person. Telephone interviews lasted between 11 to 29 minutes (Mean: 19 minutes). The face-to-face interview lasted 45 minutes.

The interview questions were exploratory and based on existing literature. The interview guide addresses four main topics: (1) willingness to self-generate health data; (2) the way they would collect and record this data; (3) identifying whether the self-generation of data causes concern, (4) willingness to share self-generated data for research and adapting their treatment or the treatment of those they care for (Appendices A and B). All participants received information sheets in person or by email (see Appendix C). Written consent was obtained from all participants who undertook face-to-face interviews (see Appendix D). Participants whose interviews were held by telephone provided oral consent (see Appendix E). All interviews were audio recorded and transcribed verbatim with participants' consent. Participants' names and other references that may lead to their identification were removed from the transcripts and names were replaced by alias for each participant. Preliminary data analysis of the interview transcripts was carried out simultaneously with data collection to promptly identify redundant information and subsequently stop sampling as necessary using the NVivo software V12. Subsequently, an in-depth analysis directed at the topics under study in this thesis was carried out drawing on deductive and inductive approaches. First, deductive analysis was undertaken based on the literature review to identify relevant themes and categories for the study. Then, inductive analysis was carried out using open, selective, and axial coding to allow the emergence of themes, categories and subcategories and to contrast these with the themes and categories identified in the previous deductive phase, to check whether the data collected revealed new information on the topic. Researcher triangulation was used to ensure the quality of the findings.

4. RESULTS

Participants' motivations to self-generate and share health data for different purposes, the concerns underlining unwillingness to do so, and the preferred technologies to engage in data self-generation are presented in this section and illustrated by direct quotes from the semi-structured interviews conducted with people affected by rare diseases, including patients and their informal carers. Participants' sociodemographic characteristics and involvement with patient organizations are described in Table 1.

Results show that most participants were willing to self-generate health data. Motivations to engage in health data self-generation are varied and include supporting and advancing biomedical research, acquiring a better understanding of disease, obtaining personalized treatments, and improving disease management. As for concerns, there were no relevant differences between patients and informal caregivers about the worries expressed. The latter included data breaches, the inability to use the tool to collect your health information, and the time spent collecting it.

The majority of participants would opt for the use of digital technologies to self-generate data, namely phone apps and computers. Only a few participants preferred to use analog technologies such as paper notebooks. This preference was motivated by the convenience and ease of collecting and storing health data.

Both patients and informal carers are more willing to share self-generated health data for personalized medicine and for biomedical research than other research purposes. In the case of the latter, most of the interviewees stated that their willingness to share would depend on the purposes for which the data would be used for.

Patients and their informal carers expressed relatively convergent views on most of the themes under analysis. For this reason, results are presented jointly for both groups. Nevertheless, the differences found between them are reported whenever they emerge.

Table 1 Participants' sociodemographic characteristics and involvement with patient organizations

Participants	Gender	Age	Education	Marital status	Occupation[1]	Involvement in patient organization
Patient 1	Male	18-29	Secondary education	Single	White-collar	No
Patient 2	Male	30-49	Secondary education	Living w/ partner	Blue-collar	No
Patient 3	Female	18-29	University education	Single	Student	No
Patient 4	Female	>49	Secondary education	Divorced	Retired	No
Patient 5	Female	30-49	Secondary education	Married	Unemployed	No
Patient 6	Male	30-49	Secondary education	Married	Blue-collar	No
Patient 7	Male	30-49	Secondary education	Single	Blue-collar	No
Patient 8	Female	30-49	Primary education	Married	Pensioner	No
Patient 9	Female	30-49	Secondary education	Single	White-collar	No
Patient 10	Female	30-49	University education	Living w/ partner	White-collar	No
Patient 11	Female	30-49	University education	Married	White-collar	No
Patient 12	Male	30-49	University education	Single	White-collar	No
Patient 13	Male	18-29	Secondary education	Single	Blue-collar	Yes
Patient 14	Female	30-49	University education	Married	White-collar	No
Patient 15	Female	18-29	Secondary education	Single	Student	No
Patient 16	Female	>49	Secondary education	Widowed	White-collar	No
Patient 17	Male	30-49	Secondary education	Single	Pensioner	No
Patient 18	Male	18-29	Secondary education	Single	Unemployed	No
Patient 19	Male	18-29	Secondary education	Single	Student	No
Patient 20	Male	18-29	Secondary education	Single	Student	No
Patient 21	Female	18-29	Secondary education	Single	Student	No
Patient 22	Female	18-29	Secondary education	Single	White-collar	No
Patient 23	Male	18-29	Secondary education	Single	Student	No
Patient 24	Female	18-29	Secondary education	Single	Blue-collar	No
Informal carer 1	Female	30-49	University education	Living w/ partner	White-collar	No
Informal carer 2	Female	30-49	University education	Married	White-collar	No
Informal carer 3	Female	30-49	University education	Living w/ partner	White-collar	No
Informal carer 4	Female	30-49	University education	Married	White-collar	No
Informal carer 5	Female	18-29	Primary education	Living w/ partner	Unemployed	No
Informal carer 6	Female	30-49	University education	Married	White-collar	No
Informal carer 7	Female	30-49	University education	Married	White-collar	Yes
Informal carer 8	Male	>49	Secondary education	Married	White-collar	Yes
Informal carer 9	Male	>49	Secondary education	Married	Blue-collar	No
Informal carer 10	Female	30-49	Secondary education	Married	White-collar	No
Informal carer 11	Female	30-49	Primary education	Married	Blue-collar	No
Informal carer 12	Male	30-49	Primary education	Married	Blue-collar	No
Informal carer 13	Male	>49	Secondary education	Married	Blue-collar	No
Informal carer 14	Male	>49	Primary education	Married	Blue-collar	No
Informal carer 15	Male	30-49	University education	Living w/ partner	White-collar	No
Informal carer 16	Male	30-49	Secondary education	Living w/ partner	White-collar	No

[1] Participants professions were omitted to reduce the risk of re-identification and replaced by the categories "white collar" and "blue collar", which denote different types of workers (for example, workers who perform administrative or managerial tasks in the case of white-collar professions and workers who engage in manual labour or skill trades such as construction or maintenance).

4.1. Perspectives about engagement with self-generating health data

Most patients and informal carers were willing to self-generate health data. However, the main motivation for informal carers to engage in health data self-generation was to help advance scientific research, while for patients the possibility of using that data to adapt and improve their treatment and disease management was a key motivation to engage.

None of the participants explicitly stated that they were unwilling to generate data. However, an in-depth analysis of the concerns evoked when considering data self-generation points to several challenges that will likely inhibit data collection should that request be made in the future. These challenges include the significant commitment needed for and burden added by daily data collection, lack of time, fears of information leakages and misuse of personal data, and a perceived incapacity to undertake data registration tasks using digital devices and software.

4.1.1. Motivations to engage in health data self-generation

Participants expressed several motivations for self-generating health data. They were primarily oriented toward contributing to research and advancing medical knowledge, gaining a better understanding of disease, and obtaining health benefits following improved disease management and access to personalized treatments. In some cases, engagement in health data self-generation was also driven by a sense of duty to get involved in research and contribute to the common good.

Patients showed a strong inclination to contribute their health data to support medical research and the development of treatments. This motivation was strongly grounded on most rare diseases being untreatable and often lacking therapies that can ease patients' symptoms and help them have a better quality of life: "I think (…) [it would be] a way of contributing my information, my data, to developing solutions to rare disease problems, mainly to obtain a cure or a treatment." (Patient 12). Similarly, another patient expressed both disappointment with learning that his disease had no cure and the hope that one would be found

in the future: "being told that there is no cure, you know? You always hope that there is, that there is a cure, you know?" (Patient 13).

Patients also highlighted the personal benefits that may derive from self-generating health data. These entailed improved disease management and access to more personalized treatments. As one patient noted, collecting data at home and sharing it with care providers could facilitate better management of his disease by reducing the number of appointments and visits to the hospital, as well as exposure to unnecessary risks (e.g. infection):

> "Because I think it's also a way for us to help. For example, I have routine appointments. I go sometimes to the hospital to do that screening and it's away from home. [So,] it's one less appointment and less risk I take at the hospital." (Patient 20)

Other patients were keen to provide their health data to professionals because they believed that would enable the tailoring of treatments to their specific needs: "(....) if we talk about my doctors, my medical team, adapting the treatment based on this (...) information." (Patient 24). In some cases, patients were especially sensitive to the need to participate in research and collaborate to advance science by sharing their self-generated data:

> "I don't know if it's because I'm in the field, or if there are a lot of people with a perspective similar to mine, because the advancement of medicine and other sciences doesn't happen without research, nor the collaboration of certain people and, in this case, if I can help make it happen, I will." (Patient 3)

Informal carers shared similar motivations, emphasizing the importance of contributing to research and promoting the discovery of new treatments, as well as enhancing their own understanding of the disease affecting their offspring:

> "(...) It ends up being a contribution to research, (...) in the end, the aim will be to find a more suitable treatment, or in the future, right? It also ends up contributing (…) [to] maybe also trying to find out what's going on. Or what happened, that led him to have the congenital problem he has, right?" (Informal carer 12)

For some carers, engaging in health data self-generation and sharing it to advance medical knowledge was perceived as a duty: "(…) I [would have] no problem contributing to these studies and disseminating information, and contributing, in fact, to the evolution of studies, of medicine. I think it's almost a duty that we have." (Informal carer 6). Other carers were driven by an aspiration to promote the common good, acknowledging both the need to gather data from various patients and the broader impact that collecting such data could have:

> "This type of record, this type of information, will always serve as parameters, for example, for studies that are useful to other people, right?" (Informal Carer 8)

> "This community spirit... there you go, I believe that for certain types of research, it's necessary to use real data from my son and other children. And, so, only the sum of all these data can create averages, can create a viable range to achieve results. So, it would be for the common good, of course." (Informal carer 15)

4.1.2. Concerns underlining unwillingness to engage in health data self-generation

Although none of the participants expressed refusal to self-generate health data, several patients and informal carers articulated concerns that hindered their willingness to engage. These concerns centered around the time and effort

required to collect data and the burden that would add to daily routines, fears related to data security and privacy breaches and potential misuse of personal data, and incapacity to undertake data registration tasks using digital technologies.

A significant barrier to data collection was the effort and time commitment involved. Both patients and carers expressed that incorporating data collection into a daily routine was cumbersome and challenging to maintain over an extended period:

> "(...) the inconvenience of having to do (...) this [data collection] daily. Do you understand? I have to take time out of my life to do that, that's all." (Patient 14)

> "As long as it wasn't a whole year... every day... Well, if I had to do that, obviously I'd have to do it, wouldn't I?" (Patient 4)

> “I don’t know if I can weigh her [daughter] every day because I work. Sometimes I come home at midnight and wake up at 6 AM to go to work.” (Informal carer 5)

> “Oh, I don’t know if I can do it if I have time for doing all those registries (…)” (Informal carer 9)

Concerns regarding the security and privacy of health data were present, especially among patients. Participants worried that the data gathered could be accessed by unauthorized parties and their privacy lost:

> " (…) if the apps would guarantee some safety and wouldn’t give access of information to everybody (…) because I think that we all know that nowadays data is not secure, neither on the computer nor in other places, (…) a hacker manages to obtain data in an extraordinary way, doesn't he?" (Patient 3)

In part, fears of data breaching were explained by lack of trust on digital data processing organisations and exposure to instances of online scamming:

> "Considering the scams that are going on online, in terms of credit cards and so on, we have to be very careful with what we trust these days (...) because if I see that the app doesn't give us confidence, I wouldn't choose that app." (Patient 7)

A patient with a background in computer science emphasized the importance of having consent procedures in place. He also cautioned to the risk of data misuse being a central barrier to health data self-generation and sharing:

> "Let's say yes [I have concerns], because we're more or less inside this area of information, because I'm studying computer science and I know about the consent we have to have. And I think the misuse of my information is the main constraint." (Patient 20)

Lack of access to digital technologies and limited digital literacy posed significant barriers to engagement with data self-generation. Some participants did not have the devices needed to gather data in a digital format:

> “(…) if I had a proper smart phone which supports those kinds of things [apps], but if I have a cheap one [phone], I can’t do it [collect data trough apps].” (Informal carer 14)

> "If I had the necessary equipment, yes, I wouldn't mind. But I don’t have the equipment." (Patient 20)

Other patients explained that they either lacked the skills to make full use of digital devices or were reluctant to use them regularly:

> "(…) because, for example, as I don't have [phone apps], and I don't know [how to use them] (...) I don't have a great ability to install [them]." (Patient 5)

"A computer, yes. A tablet, I don't, I don't use those things... I understand computers and I can handle them. When it comes to other things, I'm far, far behind." (Patient 24)

"It's like this, every day... it's not that I don't have a lot of patience, but I'm not one for spending a lot of time on my cell phone. Or the computer, or something." (Patient 1)

One informal carer stated she would not gather data using digital devices because she distrusted digital technologies overall:

“No, I wouldn’t use them [digital devices] because I don’t trust [them]. I am very suspicious (…) I don’t trust technologies.” (Informal carer 2)

4.2. Preferred technologies to self-generate health data

Most participants preferred to use digital technologies to collect their health data. Patients and informal carers expressed similar reasons for opting for digital devices including greater effectiveness in facilitating data collection and storage and expediency, namely by decreasing the odds of losing the data and by enabling them to send it to any end-user from the comfort of their homes and thus avoid unnecessary traveling to sites where they could be exposed to health risks:

“If I had to send it [self-generated data], right? In other words, I'd probably prefer [to send] it online, of course, wouldn't I? I was already doing it [collecting data using digital devices] and I'd send it, because under these conditions and as you can see, having a rare disease, I'm quite scared of it [going to the hospital] (laughs)... now I'm a bit more withdrawn at home, so I'd prefer online. Instead of going to the post office or the hospital [to hand in the data].” (Patient 10)

> “I also think that now it [using digital technologies] is the best way... isn't it? You rarely lose your data. There's always an account or something... and on paper, you can lose it and not have it. That has happened to me.” (Informal carer 5)

Participants’ preferred digital devices to self-generate data included digital apps, computers, and electronic platforms:

> "A [smartphone] app would be ideal, because the information would be available straight away and we would be able to enter it easily, for both parties”. (Informal carer 3)

> "Yes, any [smartphone] app. Today it's very simple to make an app and see this data with the app." (Informal carer 16)

> "We usually record everything online now, so on Excel-type platforms." (Patient 22)

Participants' preference for the use of analog technologies was grounded on a lack of access to suitable digital devices or limited knowledge of how to use them.

> "My aunt doesn't have access to a computer (...). It would be easier for her to do it [collect data] on paper, wouldn't it?" (Informal Carer 4)

> "Ah, yes, yes. Then of course I wrote it [health information] down in a notebook. I wrote everything down in a notebook and forget the computers because I couldn’t understand it. It's all in a notebook and then I'd make photocopies." (Informal carer 1)

A few participants stated that they had no preference for a specific technology to gather data and that both digital and analog technologies would be plausible options:

> "Oh, well, at the moment I think I'd prefer... it doesn't matter, at the moment it doesn't matter to me. Paper, or Excel (...) there's a whole security system around Excel now too. If it was Excel, around the computer there's a whole security system, but... sometimes it's a lot, sometimes it's not much, but there's always some security. But at the moment it doesn't matter to me whether it's in Excel or on paper." (Patient 3)

4.3. Perspectives about sharing self-generated health data

Three main contexts were identified in which motivations and concerns related to health data sharing were investigated. The majority of participants, both patients and caregivers, expressed a willingness to share their data for personalized medicine and biomedical research. The primary motivations were the possibility of obtaining treatments tailored to their specific health conditions and the prospect of developing new drugs, new treatments, and improving their quality of life.

Regarding other types of studies, participants emphasized the importance of knowing the purpose of the research and the institution responsible for data analysis before deciding. A clear distinction was observed in participants' willingness to share their data with pharmaceutical industries and health insurance companies. While most were open to sharing data with pharmaceutical companies due to their role in developing new drugs, the same willingness was not observed regarding health insurance companies. Participants were more inclined to share data with insurance companies only for demographic research purposes.

Participants' main concerns were related to data breaches and misuse, with consequences such as discrimination.

4.3.1. Sharing self-generated data for personalized medicine

All the participants stated that they were willing to share their health data for the purpose of adapting treatments to their specific needs. Participants were motivated by the added value this would bring to patients, both by improving their treatments and their quality of life. The concerns mentioned involved the confidentiality of the data shared, the need to act responsibly when using that data, and the difficulties involved in adapting the treatment for patients' needs.

4.3.1.1 Motivations to share self-generated data for personalized medicine

Participants saw considerable value in sharing health data to tailor treatments to meet the specificities of their health conditions. They compared it to previous historical advancements in medicine, emphasizing that sharing relevant health information could lead to better treatment outcomes and improved quality of life.

> "I see no inconvenience [in sharing data]. In fact, it's a benefit for him [son]. It's always the goal to help. And I compare his illness a bit to diabetes. Diabetes was a mar issue until chemical insulin was discovered, right? Producing it in the lab helped and facilitated life for the population." (Informal carer 3)

> "We never have everything [health indicators] the same, right? When we do some tests or exams, it's never exactly the same. Some things improve, and some things might not be as good as before, so it's always good to share [health data]." (Patient 2)

"Ah, [the benefit of sharing data is] improving my quality of life, my health. Because I have a young daughter, and I think a lot about this. (…) Because unfortunately, I have some health problems, and I think a lot about being here for my daughter and my husband." (Patient 5)

Participants recognized that sharing self-generated health data also improved disease management by helping patients both to be informed of changes in their health conditions and by ensuring that treatments is can be adjusted when needed so as to remain responsive and effective.

"Because I think it was a way for people to know... what to take care of in their health, to check [metabolic blood] levels... compare levels with others. (…) It helps to try to save me from getting ill or help treat a disease..." (Patient 22)

"For example, we have consultations once a year. During the year, a lot can happen, and if someone is undergoing a type of treatment with better control, they might adjust the medication before the next scheduled consultation." (Patient 20)

"To adapt [treatment], well, not only for her (daughter] but also for other cases, right? And to be more helpful with this [adapting treatments], there will always be the need to know what's happening with her." (Informal carer 5)

Trust in medical teams is fundamental when it comes to tailoring treatments for rare and complex conditions. Several patients expressed confidence in their healthcare providers to make necessary adjustments based on the specific details of their conditions, which entailed data collection over time. As a patient explained: " As you understand, I have a rare disease that few people know about.

So, at São João [hospital], the only people I trust absolutely to adjust or make any changes to my treatment are my medical team.” (Patient 24)

Patients and carers recognized that treatments should be personalized, taking into account individual variations and specific characteristics. They believed that increased access to and a better understanding of each patient’s health data can lead to more effective and personalized treatment approaches:

"I think each person is unique, and we're always... well, medications are the same for everyone, but... people might have the same problem but different needs, and based on their weight or blood pressure, they might react better to one type of treatment than another... so, I think these are always important data points when making important treatment decisions." (Patient 15)

4.3.1.2 Concerns about sharing self-generated data for personalized medicine

The interviewees highlighted some concerns about data confidentiality and the challenges of adapting to new treatments. They emphasized the importance of privacy and the need for clear information about how their data would be used. Some participants were wary about how their data would be kept confidential and the conditions under which it would be shared. They wanted assurances that their data would be used responsibly and anonymously to protect their identity:

"That [sharing self-generated data] would be something to consider, as I said earlier, in terms of data confidentiality. And in this case, the approach to data sharing. I would have to know for what purposes the sharing [of data] would be and then, yes, I would look into things." (Patient 7)

"One of the things I might ask for is her anonymity, in the sense of not presenting, her face there (…) Because she will grow up and then she will understand, and maybe I don't know to what extent she will one day say: 'oh mom, my face is there!' or 'my name is there'. But the name I am still okay

with, now the face I don't think so. I would have to consider it" (Informal Carer 11)

Participants also voiced concerns about the practical challenges and strict routines required when adapting to new treatments hinting on the downside of sharing self-generated data for the purposes of tailoring treatments.

“As long as they [health professionals] didn't change too much [by accessing and assessing self-generated data], because that's the way it is with these kids, the worst thing is changing things. I think the more you change it, the worse it gets.” (Informal carer 7)

4.3.2. Sharing self-generated data for biomedical research

All participants were willing to share their self-generated data for biomedical research, although some patients made that contingent on the end goal of research and were keener to share if the data was used to advance research on their disease. Other motivations to share included the possibility of discovering new treatments and cures, increasing quality of life, and supporting research that could benefit people with other health conditions.

Both patients and carers expressed concerns about sharing the data, namely potential breaches of privacy and subsequent data misuse, including public disclosure and undesirable labeling that could lead to discrimination. They also pointed to knowing and trusting research teams as a central aspect in reducing the impact of those concerns.

4.3.2.1 Motivations to self-generate data for biomedical research

Most participants acknowledged the importance of contributing self-generated health data to scientific studies that aim to improve health outcomes for themselves and for future generations. Participants were motivated to do so by the potential benefits they believe can derive from sharing their data research, namely the identification of new diseases and improved diagnosis, the discovery

of new cures, and the development of more effective treatments. This sentiment drovn their desire to support ongoing research and advance medical knowledge.

"From the perspective (…) that one day in the future it [self-generated data] might help to contribute in some way... because if no one helps to obtain data, we will never be able to help or find, not the cure, but something that helps to improve the quality of life, right? That's why I don't mind contributing." (Informal carer 3)

"To help in the development and research and discovery of (…) new cures, new treatments... Yes, not only for personal use, but also for the future, right? The more we know, and the more we study, the more development there is, the more chances there are for the future, right?" (Informal carer 2)

"Scientific research produces advances in medicine and produces advances in science that will then, in principle, produce better treatments, right? And a more appropriate treatment for each pathology, let's say, isn't it? And I think that's a good goal, isn't it? Because research, if research isn't done, there's no advance in medicine, there's no advance in treatments. If it is done, maybe in the future there will be things that can be prevented earlier or there will be treatments that will be more appropriate than those that exist today". (Informal carer 10)

Participants also expressed a strong sense of duty to help others through the sharing of their health data. Some referred to the importance of expanding existing databases to enable new types of scientific inquiry and enhance the outcomes of future research.

"This is relatively political, but I advocate for a society that is democratic and has principles of equality, and therefore... I believe in this duty of community

support and that sharing my data can save the lives of my peers, whether they are close to me or not." (Informal carer 15)

"These are always data that can help future generations, or that can create a database that leads to other types of developments for students. I mean, in the end, we need to be available to help others whom we don't even know if we are going to help or not, right?" (Informal carer 12)

Some patients expressed willingness to share their self-generated data for biomedical research particularly if it focused on their own health conditions and could benefiting them more directly.

"[I would share my data] with the hope, right, that they would investigate, maybe, I don't say [to find] a cure, because maybe there isn't one, right? But something that improves our quality of life, isn't it? For my disease... in other words, what I would like is for there to be some research into [name of disease], you know, my specific disease." (Patient 10)

"[I would share my data] mainly to better understand my own health, right? That would certainly help at the level of scientific research too." (Patient 5)

4.3.2.2 Concerns about self-generating data for biomedical research

Participants showed concern about sharing their health data for biomedical research related to potential breaches to privacy and data misuse, including the risk of discrimination. Knowing and trusting research teams emerged as safeguard into easing these concerns.

Participants emphasized the importance of guaranteeing anonymity and maintaining confidentiality when sharing their self-generated health data and ensuring that their personal information remains secure. There was also a strong

emphasis on the need for professional secrecy and the careful handling of personal data.

"I think [the data can be protected with)] with numbers, there's no need to expose the person's name. You understand? Who will know that the blood of 7881 belongs to... I think with numbers, there's no need to expose the person's name. Right? Professional confidentiality." (Informal carer 4)

"Total privacy [of the data shared]. In other words, everything I know about you and everything you know about me would remain just between us, you know?" (Patient 7)

Participants feared that their data could be mismanaged, unduly disclosed to the public and used in inappropriate ways with potentially negative consequences. They were also afraid that their information could be used to judge and label them in ways conducive to discrimination.

"If this information is shared and it is not well used or not well managed by the scientific committee, not well managed in the sense that it is shared without concrete results, or it is released to the public, or it is disseminated without having anything concrete. In that case, I think it's a bit of a lack of responsibility" (Informal carer 10)

"I assume there is always confidentiality in the information, that the data is handled with care, in such a way that there is no public disclosure unless for the research itself, right?" (Patient 11)

"I do make a distinction with DNA. Therefore, those who have nothing to fear have nothing to hide. But I do worry about the misuse for dubious scientific

purposes... like cloning, which doesn't exist yet but could one day." (Informal carer 15)

"Maybe because I wouldn't want to be labeled, you understand? I think there's no need to have a label: person X shared this clinical information or this... I think there was no need for that. Because if it's negative it could be censored, there could be some reason for censorship. " (Informal carer 14)

Participants expressed a desire to know the research team involved in the study. They felt that having a clear understanding of who is conducting the research and their intentions would increase their trust and willingness to share their data.

"(…) I don't give it [self-generated data] just like that. Because they say: 'Can we do an analysis for research with your blood?' Just out of the blue, no [they don't request data that way]. I need to know things [research purposes] properly because it's my blood, it's my DNA. (Patient 24)

4.3.3. Sharing self-generated data for other research purposes

The interviewees were also asked about sharing self-generated health data for research purposes other than personalized medicine and biomedical research. Research conducted by the pharmaceutical industry and health insurance companies were provided as examples of these other purposes.

Most of the participants stated that their willingness to share data was dependent on the purposes of the studies and the institution hosting them. Supporting research that could benefit other people, for example through the development of medical devices, and the importance of contributing data for population or disease statistics were key motivations to share.

Only a few participants expressed unwillingness to share data for research conducted outside academic and health care institutions, with patients being slightly less willing to share than carers.

Primary concerns included the use of data for purposes other than the patient's well-being, with participants feeling health insurance companies are profit-driven and may not offer any tangible benefits to improve patient's lives. This perception was also held regarding the pharmaceutical industry though to a much lesser degree as participants stated the importance of its role in developing new medical drugs.

4.3.3.1 Motivations to share self-generated data for other research purposes

Most participants stated that they would consider sharing self-generated data for purposes other than biomedical research and personalized medicine. However, their willingness to share was often contingent on the specific purposes of the study and the type of institution leading them. Some participants emphasized the need for being provided with clear information about the studies' goals, their duration, and how their data would be used to make an informed decision:

> "Ah, I don't know [if I would share data]. I would need to know what it [data] is for. (…) I would only really make these decisions: 'look, yes, I participate'. I would only say [that if] (…), this [scenario] is a bit hypothetical and I think that, in general, there is no doubt that I think we should contribute everything [data], but then [to share] I would want all the information: what is it for? What is this study for? How long will it last? Where will it be published? And what are the objectives? This information would be fundamental for me to decide everything." (Informal carer 6)

"Well, it depends on the main goal of the [study], but… because [for] insurance companies, I don't know [if I would share]. (…) But for a pharmaceutical company, yes. Because they are the ones who make... it's the whole industry that makes antibiotics, right?" (Patient 23)

Participants made a distinction between sharing health data with pharmaceutical companies and health insurance companies. While the former was perceived to be involved in the development of medicines with direct benefits to patients, the purposes of the latter were more dubious. Some participants stated they would share data with health insurance companies provided they were transparent about their goals, while others dismissed them as potential data recipients as shown in the next sub-section.

"Yes, I think I don't see any problem with that. I don't know exactly why an [health] insurance company would need patient information for, but I think that if everything is explained... Precisely to gain knowledge... if it's not for health purposes, maybe, I don't know, for other purposes, right? No one does research without having an end in sight. So, if the end goal is to help people, even if it's not for diseases but for better quality of life, or whatever, I don't see any problem with that." (Patient 22)

Research focusing on population statistics and the making of medical devices was also referenced as addressing relevant goals and worthy of participants' data donations.

"Yes, for example, I don't know, [health] insurance companies I would say: 'maybe not'. But... well, even if it's for demographic studies or statistics or... things like that, it's always useful to have [data]. And the

more samples they have, the more accurate it will be, more effective. So, yes [I would share for that]." (Patient 15)

"If it were, for example... let's imagine, in the field of physical engineering. (...) there are people in physical engineering who specialize a bit more in the area... not in the area of medicine, but in developing devices for medicine. Yes, I wouldn't mind [sharing data for that] either, for example." (Patient 13)

4.3.3.2. Concerns about sharing self-generated data for other research purposes

Several participants expressed reservations about sharing self-generated data for research outside academic and healthcare institutions. Their concerns derived from mostly from uncertainty about whether that research would produce any benefits for data donors and the community at large. In some instances, this motivated unwillingness to share data for non-medical research:

"I would have to see, right? With whom I was sharing [the data with] and (pause)... I don't think there would be any harm, I don't know. Just seeing who it was coming from, if not, to know who I was dealing with because sometimes there are cases that (…) they want to know, but then it's not to help anyone and it's just to have our data and all those things, so no [I would not share for that]." (Informal carer 5)

"I would only share if it was specifically for medical cases where I thought it could help someone. With insurance companies or similar entities, I don't think it would bring any benefit." (Patient 21)

Participants were especially wary about sharing their health data with health insurance companies. They feared that these organizations might prioritize profit over their well-being and they questioned whether their data would truly be used to benefit their health.

"I don't see the point of the health] insurance company having my weight and other personal information when their goal is to insure me, not my characteristics. They don't need to know my specifics to provide health insurance. I think it would just be a way for them to further exploit the market and increase their profits." (Patient 23)

"Because I think not [I would not share data with], [health] insurance companies, if I'm honest. They only want money." (Patient 13)

Some participants also expressed concerns about sharing their data with pharmaceutical companies. Unlike others who acknowledge the potential benefits deriving from pharmaceutical research (e.g. discovery of new drugs), the following participants doubted of their intentions to improve patients' health: were also noted, with some participants questioning the benefits of data sharing in this context.

> "While I believe that well-directed scientific research in medicine could lead to improvements for other patients in the future, I think the pharmaceutical industry is more concerned with financial profit than with improving patient outcomes." (Informal carer 10)

> Will what we're adjusting [treatment] do any good or harm? That's up to her. They [pharmaceutical industries] are thinking of earning more [money] (Patient 9)

5. DISCUSSION

To our knowledge, this is the first study to examine the views of individuals with rare diseases, and their informal carers, about engagement with generating and sharing patient health data for personalized medicine and biomedical research in Portugal. Health digitization and the expansion of data-intensive research can afford numerous public health benefits, including disease prediction and prevention, improved care management, and the discovery of new treatments and cures that can significantly change the well-being and quality of life of people with chronic conditions (Abernethy et al., 2022; Benson, 2016; Flores et al., 2013). To achieve these goals, and to distribute their benefits equitably, it is necessary to gather data from across diverse population subsets (Atutornu et al., 2022; de Freitas et al., 2021; Sirugo et al., 2019). This study aims to contribute timely evidence about public and patient perspectives on health data self-generation and sharing that can be used to develop inclusive research recruitment strategies and data governance practices with particular attention to people affected by rare diseases.

The results show that most of the rare disease patients and informal carers interviewed are willing to self-generate health data, although they expressed different motivations to do so. While most informal carers express willingness to engage based on a perceived need to carer contribute to advance scientific research and to gain a better understanding of the disease, patients were more often motivated by the possibility of accessing personalized treatments and improving their disease management. This is in line with existing studies, which highlight a growing trend for patient engagement in health data collection as a way of promoting personalized healthcare and improving patient outcomes (Austin et al., 2020; Öberg et al., 2018; Papa et al., 2020; Shen et al., 2022). Patients leaning toward intrinsic motivations for involvement (i.e. delivering personal benefits) (Simmons & Birchall, 2005), such as benefiting from personalized care and improved disease management, is not an unexpected finding. A similar disposition has been observed among people with other health conditions (e.g. mental health issues) that can be as incapacitating, which are as incapacitating, stigma-inducing, and challenging to social acceptance and integration as are rare diseases (de Freitas, 2015). Supporting patients in

understanding how, and to what extent, they can attain those individual goals will likely help to sustain engagement in data self-generation. For example, while genomic research carries the substantial potential to deliver personalized treatments, its translation into treatments tailored to rare disease patients' specific needs has been slow (Bittles, 2019). Perhaps informal carers are more accurately informed of the current results of biomedical research, building their willingness to self-generate health data around the need to continue to bring scientific research forward. Sharing this information with patients will be important to prevent thwarted expectations and to guide their efforts toward feasible outcomes. Participants expressed concerns with health data self-generation driven by a lack of material resources and perceived limited safety guarantees. The latter concerns derive from fear of information leakages and possible misuse of personal data with negative consequences to individual privacy as shown in previous studies (Brown et al., 2022; Chen, 2020; Öberg et al., 2018). The former concerns centered around lack of time due to family and work commitments and difficulties incorporating data collection into daily routines that are rather demanding in light of the attention and care needed by people affected by rare diseases. Some participants also experienced limited digital literacy and a lack of access to digital devices, which reduced their ability to self-generate data. These barriers to engagement in data self-generation can cause those least resourced to become under-represented in research and potentially widen inequalities in access to its future benefits. Overcoming these problems requires different types of actions. Health data processing organizations, including research and healthcare institutions, need to develop patient-centered data governance policies and practices to reduce the barriers encountered and to increase the engagement of patients and their informal carers in the collection and sharing of health data. First, it is crucial to address their privacy concerns and build trust among data donors. In addition to implementing robust security measures to protect patient data, data processing organizations must prioritize transparent communication about data collection, storage, and use. Since this often entails complex information, it is advisable to involve patients and their representatives in co-developing communication strategies and educational materials so they can be conveyed using plain language and meet the needs of their end-users (Falcão et al., 2023). It is also important to promote the engagement of patients and

informal carers whose social, economic, and health conditions may be less favorable to participation. Approaching potential participants directly (rather than posting adds inviting participation) and asking how their engagement can be supported (e.g. offering access to devices and training) is a fruitful strategy to be a productive strategy to involve groups at risk of marginalization (de Freitas & Martin, 2015). This study also revealed a preference for the use of digital devices among patients and informal carers for self-generating health data. This preference was driven by factors such as ease of use, avoiding unnecessary travel to send the collected data, and the ability to store and manage data easily, and it is corroborated by literature highlighting the benefits of integrating digital health technologies into patient care, including enhanced accuracy, efficiency, and patient engagement (Antonio et al., 2022; Lavallee et al., 2020). While a subset of participants stated that they were flexible regarding indifferent to the type of technologies used to self-generate data, a few patients and carers expressed a clear preference for using analog technologies. This was motivated by a lack of access to digital devices or an inability to use them. This highlights the need for inclusive approaches that accommodate the different levels of technological proficiency among patients and carers, especially those with more difficulties, whether cognitive, financial, or lack of family support (Shan et al., 2019; Slevin et al., 2019). Health data processing organizations should offer training and support to help patients and carers feel more comfortable using digital devices.

Participants' willingness to share self-generated health data varied according to the purposes of the research. They were more inclined to share their data for biomedical research and personalized medicine than for other research purposes. They reasoned that the former would more likely derive direct benefits in terms of improving the management and understanding of their disease, and even the possibility of discovering new treatments, than in other types of research. These results align with those of a study conducted by Peng et al. (2016) who found that participants' main motivations to engage were the opportunity to better understand the disease and receive updates on related research. This points to the importance of gathering evidence on whether a return of research results would act as an incentive to engage in health data self-

generation among research participants in Portugal and what the most appropriate format to share those results would be (Lewis et al., 2021; Milne et al., 2022). Furthermore, it also draws attention to the need to communicate what type of results may be expected from studies to prevent misleading aspirations and disappointment.

Although most participants were willing to share their self-generated health data for biomedical research and personalized medicine, they expressed several concerns that needed to be addressed to ensure data sharing for research conducted outside academic and healthcare institutions (e.g. by health insurance companies or the pharmaceutical industry). A perceived lack of control over who would use their data and the fear of losing privacy and being discriminated against should data be accessed by unauthorized third parties were among the most relevant concerns. When considering sharing data for other research purposes the credibility of the institution conducting the research, its potential to generate tangible benefits for participants, and the ability to ensure the safety of their data were considered critical factors to make a decision. Participants emphasized the importance of maintaining anonymity and being explained the study's exact purposes. As some studies show, rare diseases are multisystemic that can cause severe impairment and lead to stigmatization if data is leaked and misappropriated (Brown et al., 2022; McCormarck et al, 2016). Preventing data leakages is imperative to ensure the trustworthiness of research institutions as is the setting up of redress mechanisms that clarify which type of compensation are data donors entitled to should they ever occur (Ursin et al., 2020). The credibility of research institutions is also largely dependent on data governance practices that ensure transparency and guarantee respect for data donors' rights, including that of making informed decisions about whether to share their data (Nwebonyi et al., 2022). As stated by a participant, enforcing data consent producers aligned with participants' needs and preferences is vital. While dynamic consent would likely provide valuable opportunities to share information with potential data donors about the purposes of research for which their data may be used, the fact that it depends on access to digital platforms carries disadvantages for donors who are not able or unwilling to use digital technologies (Riso & Águas, 2022). Meta consent (Ploug & Holm, 2015) may thus be a more inclusive and viable

option. It allows participants to choose between different modalities of consent, i.e. broad consent (where donors are asked to provide consent for research purposes that may not yet be entirely specified), specific consent (where consent is given for a specific purpose in one point in time), and dynamic consent (enables donors to define and modify consent preferences over time) as they receive requests to use their shared data (Ibid). Offering an option for meta-consent can contribute to fostering ongoing communication between data donors and researchers (i.e. through dynamic consent procedures) and, at the same time, ensure that participants who have a preference for analog consent technologies are also able to make informed decisions (i.e. through broad or specific consent procedures).

This study has some limitations. This study was conducted in one hospital and there is a possibility that people with fewer resources, whether material, emotional or health-related, may have refused to take part in the study. Another limitation of the study was that it did not explore which digital tools could be used to collect health data, such as whether there would be a difference in availability and barriers encountered by the participants between a device that automatically collects health data and one in which the participant must enter the data manually.

6. CONCLUSION

This thesis highlights the importance of understanding the diverse motivations, preferences, and concerns of rare disease patients and their informal carers regarding collecting and sharing health data. By addressing the barriers identified and harnessing preferences for digital devices, healthcare systems can better support patient engagement and contribute to advancing personalized medicine and biomedical research.

The insights gained from this study can guide the development of more effective health data governance frameworks and digital health interventions. For example, simplifying data collection processes through apps that automate parts of the recording can make the process more user-friendly and integrating data collection with daily activities and utilizing existing devices, such as smartphones, can help reduce the additional time required. Additionally, ensuring that the security systems used for data collection are robust will enhance patient and caregiver confidence in sharing their health data.

Future research should focus on exploring the long-term impacts of health data generated by patients and their informal carers on the health outcomes of rare disease patients. In addition, studies could investigate the effectiveness of various digital devices in different patient populations, such as automated or manual data collection devices. Understanding the factors that influence the adoption and sustained use of digital health technologies will be key to developing interventions that are effective and widely accepted.

More research is also needed to explore the impact of cultural, social and economic factors on the willingness of patients and informal carers to generate and share health data. These factors can significantly influence attitudes and behaviors and understanding them can help in the development of more targeted and effective interventions.

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APPENDIX A: Patient telephone interview guide

Guião de entrevista por telefone com doentes

(versão abreviada referente ao atual estudo)

(1) Parte introdutória: Apresentar o objetivo do estudo; dar oportunidade para colocar questões; agradecer disponibilidade para participar; pedir consentimento verbal.

(2) Recolha de informação sociodemográfica: idade, país de origem, estatuto marital, grau de escolaridade, profissão, envolvimento associativo (já fez/faz parte de uma associação de doentes?).

II. Participação individual: geração, recolha e uso de informação de saúde
Para o desenvolvimento de tratamentos médicos personalizados, tendo em conta as características individuais de cada doente, poderá ser necessária a recolha de informação de saúde do doente.
1. Se lhe pedissem para recolher as suas informações de saúde (ex. registo do peso, pressão arterial), estaria disponível para o fazer? 1.1 Se disponível, porquê? 1.2 Se não disponível: haveria outra pessoa que pudesse estar disponível para o fazer? Porquê?
2. Como poderia registar as suas informações de saúde? Que dispositivos poderiam ser utilizados? 2.1 Se lhe pedissem para as registar utilizando novas tecnologias (por exemplo, aplicação no telemóvel, tablet ou computador) estaria igualmente disponível para o fazer? Porquê?
3. Há algum aspeto relacionado com a recolha das suas informações de saúde que lhe cause preocupação? Qual? Porquê?
4. Partilharia as informações que recolheu com a finalidade de adaptar o tratamento às suas caraterísticas e necessidades? Porquê?
5. Partilharia as informações que recolheu para efeitos de investigação sobre a sua doença? Porquê? 5.1. E se a investigação se centrasse noutra doença, partilharia a informação que recolheu para esse efeito? Porquê? 5.2 E se a investigação não estivesse relacionada com o campo da medicina, partilharia a informação para esse fim? Porquê?
II. Finalização
6. Há algum aspeto que queira partilhar e que eu não tenha perguntado?
7. Tem alguma questão que gostaria de colocar?

APPENDIX B: Informal carer face-to-face interview guide

Guião de entrevista face a face com cuidadores informais

(versão abreviada referente ao atual estudo)

(1) **Parte introdutória:** apresentar o objetivo do estudo (folha de apresentação do estudo); consentimento informado; dar oportunidade para colocar questões; agradecer disponibilidade para participar.

(2) **Recolha (áudio) de informação sociodemográfica:** idade, país de origem, estatuto marital, grau de escolaridade, profissão, envolvimento associativo (já teve alguma experiência enquanto membro de associações de doentes?).

I. Participação individual: geração, recolha e uso de informação de saúde
Para o desenvolvimento de tratamentos médicos personalizados, tendo em conta as características individuais de cada doente, poderá ser necessária a recolha de informação de saúde do doente. 1. Se lhe pedissem para recolher as informações de saúde da pessoa de quem cuida (ex. registo do peso, pressão arterial), estaria disponível para o fazer? 1.1 Se disponível, porquê? E na sua ausência, haveria outra pessoa que pudesse fazer a recolha? 1.2 Se não disponível: haveria outra pessoa pudesse estar disponível para o fazer? Porquê? 2. Como poderia registar as informações de saúde da pessoa de quem cuida? Que dispositivos poderiam ser utilizados? 2.1 Se lhe pedissem para as registar utilizando novas tecnologias (por exemplo, aplicação no telemóvel, tablet ou computador) estaria igualmente disponível para o fazer? Porquê? 3. Há algum aspeto relacionado com a recolha das informações de saúde da pessoa de quem cuida que lhe cause preocupação? Qual? Porquê? 4. Partilharia as informações que recolheu com a finalidade de adaptar o tratamento às caraterísticas específicas da pessoa de quem cuida? Porquê? 5. Partilharia as informações que recolheu para efeitos de investigação sobre a doença da pessoa de quem cuida? Porquê? 5.1 E se a investigação se centrasse noutra doença, partilharia a informação que recolheu para esse efeito? Porquê? 5.2 E se a investigação não estivesse relacionada com o campo da medicina, partilharia a informação que recolheu para esse fim? Porquê?
II. Finalização
6. Há algum aspeto que queira partilhar e que eu não tenha perguntado? 7. Tem alguma questão que gostaria de colocar?

APPENDIX C: Information sheet

Informação sobre o estudo

Estamos a desenvolver um estudo sobre as opiniões de doentes, cuidadores informais, profissionais de saúde, gestores, técnicos e pessoal administrativo quanto aos benefícios e riscos associados à partilha de informação de saúde e à participação dos cidadãos na sua governação, no âmbito das doenças raras.

Gostaríamos de contar com a sua participação.

Antes de decidir, é importante que saiba mais acerca deste estudo e do que lhe é pedido se aceitar participar.

Por favor, leia atentamente este folheto informativo e coloque todas as perguntas que achar necessário.

Por que queremos falar consigo?

A finalidade deste estudo é perceber de que forma as necessidades, preferências e valores das pessoas com doenças raras, cuidadores informais, profissionais de saúde, gestores, técnicos e pessoal administrativo podem ser incluídas na governação de informação de saúde.

Quando falamos de informação de saúde estamos a referir-nos a todo o tipo de informação direta ou indiretamente ligada à saúde de uma pessoa e à sua história clínica e familiar (por exemplo, registos clínicos, resultados de análises e outros exames complementares, intervenções e diagnósticos).

Serão convidadas a participar pessoas com mais de 18 anos de idade seguidas nos centros de referência para as Doenças Hereditárias do Metabolismo e de Cardiopatias Congénitas do Centro Hospitalar Universitário de São João (CHUSJ), e seus cuidadores informais, assim como profissionais de saúde, gestores, técnicos e pessoal administrativo da equipa alargada.

Em que consiste a sua participação?

Gostaríamos que participasse numa entrevista a realizar num local reservado do CHUSJ, ou numa localização à sua escolha, com uma duração de 30-60 minutos. Estamos disponíveis para esclarecer todas as dúvidas e questões que possam surgir.

Quais serão os benefícios da sua participação?

Será participante de um estudo inovador que procura compreender os benefícios e os riscos associados ao envolvimento dos cidadãos na governação de informação de saúde, contribuindo para:
- Identificar os desafios inerentes à partilha de informação no âmbito das doenças raras e seus benefícios;
- Incentivar o debate em torno das respostas aos desafios que enfrenta a proteção de informação de saúde;
- Adaptar a governação de informação de saúde às necessidades, preferências e valores de todas as pessoas envolvidas.

A informação é confidencial?

Sim, nos termos exigidos pela lei. Toda a informação que partilhar connosco será vista somente pelos membros da equipa de investigação.
A informação será armazenada de forma segura. Sempre que as informações recolhidas forem utilizadas, nunca será usado o seu verdadeiro nome.

Sou obrigado/a a participar?

Não. Caso decida não participar, esta decisão não terá quaisquer desvantagens nem influenciará os cuidados de saúde. Mesmo depois de aceitar, poderá desistir em qualquer altura e sem justificação.

Quem coordena o estudo?

A coordenação é da responsabilidade do Instituto de Saúde Pública da Universidade do Porto (ISPUP), sendo a investigadora principal Cláudia de Freitas.

Este estudo é financiado pela **Fundação para a Ciência e a Tecnologia.**

Como será usada a informação?

Os resultados deste estudo serão divulgados de diversas formas (relatórios, artigos científicos e comunicações orais), junto da comunidade científica e de pessoas que podem tomar decisões em relação às políticas que regulam a proteção de dados de saúde.

Obrigado por ter lido este folheto!

A sua participação será muito valiosa.

A aplicação do questionário apenas prosseguirá depois de colocadas todas as questões pelo participante e após assinatura do consentimento informado.

Ser-lhe-á dado este folheto informativo e uma cópia do consentimento informado.

Para qualquer dúvida, sugestão ou comentário, por favor entre em contacto connosco:

Investigadoras:

Cláudia de Freitas | Susana Silva

Instituto de Saúde Pública
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ISPUP
INSTITUTO DE SAÚDE PÚBLICA DA UNIVERSIDADE DO PORTO

FOLHETO DE INFORMAÇÃO AO PARTICIPANTE

Participação dos cidadãos na governação de informação de saúde: partilha e proteção de dados em doenças raras

APPENDIX D: Consent DATAGov - face-to-face semi-structured interviews

CONSENTIMENTO INFORMADO, LIVRE E ESCLARECIDO PARA PARTICIPAÇÃO EM INVESTIGAÇÃO

de acordo com a Declaração de Helsínquia e a Convenção de Oviedo

Designação do Projeto: Participação dos cidadãos na governação de informação de saúde: partilha e proteção de dados em doenças raras (DATAGov)

Por favor, leia com atenção a seguinte informação. Se considerar que algo está incorreto ou pouco claro, não hesite em solicitar informações adicionais. Se concordar com a proposta que lhe foi feita, por favor assine este documento.

O Projeto de Investigação *Participação dos cidadãos na governação de informação de saúde: partilha e proteção de dados em doenças raras (DATAGov)* pretende conhecer as opiniões de diversas pessoas, incluindo doentes, cuidadores informais, profissionais de saúde, gestores, técnicos e pessoal administrativo quanto aos benefícios e riscos associados à partilha de informação de saúde e à participação dos cidadãos na sua governação, no âmbito das doenças raras.

Sei que a participação nesta fase do estudo consiste na realização de uma entrevista, realizada por um(a) investigador(a) e com uma duração entre 30 e 60 minutos.

Foi-me garantido que todos os dados relativos à identificação dos participantes neste estudo são confidenciais e que será mantido o anonimato. Além disso, sei que posso recusar-me participar ou interromper a qualquer momento a participação no estudo, sem nenhum tipo de penalização por este facto. Foram-me fornecidos os contactos da equipa de investigação, no caso de sentir necessidade de os usar posteriormente. Foi-me dado todo o tempo de que necessitei para refletir sobre esta proposta de participação e foi-me dada a oportunidade de fazer as perguntas que julguei necessárias, tendo obtido resposta satisfatória.

Declaro ter lido e compreendido este documento, bem como as informações verbais que me foram fornecidas. Desta forma, aceito participar de livre vontade neste estudo de acordo com os itens assinalados em baixo, confiando que apenas serão utilizados para esta investigação e na garantia de confidencialidade que me é dada pelo/a investigador/a. Também autorizo a divulgação dos resultados obtidos no meio científico, garantindo o anonimato.

	Sim	**Não**
Aceito participar numa entrevista qualitativa no âmbito do Projeto DATAGov.	☐	☐
Autorizo a gravação e transcrição da entrevista, que serão destruídas após a conclusão do estudo e nos prazos legalmente estabelecidos.	☐	☐
A transcrição da entrevista só poderá estar disponível para acesso pela equipa de investigação.	☐	☐

Data: ___/___/ 20____ **Investigadora responsável:** 

(Cláudia de Freitas)

Participante:

Nome:_____

Assinatura:_____

Investigador:

Nome:_____

Assinatura:_____

ID ☐☐☐☐☐

APPENDIX E: Consent DATAGov - telephone semi-structured interviews

Consentimento informado para a realização de entrevista por telefone

O Projeto de Investigação DATAGOV - pretende conhecer as opiniões de doentes, cuidadores informais e profissionais de saúde quanto aos benefícios e riscos associados à partilha de informação de saúde e à participação dos cidadãos na sua governação, no âmbito das doenças raras.

A sua participação nesta fase do estudo consiste na realização de uma entrevista telefónica com a duração aproximada de 15 a 20 minutos. Todos os dados que fornecer são confidenciais e será mantido o anonimato.

Para obter o seu consentimento, gostaria que me respondesse sim ou não às seguintes afirmações:

	Sim	**Não**
Aceito participar numa entrevista qualitativa no âmbito do Projeto DATAGov.	☐	☐
Autorizo a gravação e transcrição da entrevista, que serão destruídas após a conclusão do estudo e nos prazos legalmente estabelecidos.	☐	☐
A transcrição da entrevista só poderá estar disponível para acesso pela equipa de investigação.	☐	☐